myostatin-related muscle hypertrophy

Myostatin-related muscle hypertrophy is a rare condition characterized by reduced body fat and increased muscle size. Affected individuals have up to twice the usual amount of muscle mass in their bodies. They also tend to have increased muscle strength. Myostatin-related muscle hypertrophy is not known to cause any medical problems, and affected individuals are intellectually normal.

Frequency

The prevalence of this condition is unknown.

Genetic Changes

Mutations in the *MSTN* gene cause myostatin-related muscle hypertrophy. The *MSTN* gene provides instructions for making a protein called myostatin, which is active in muscles used for movement (skeletal muscles) both before and after birth. This protein normally restrains muscle growth, ensuring that muscles do not grow too large. Mutations that reduce the production of functional myostatin lead to an overgrowth of muscle tissue.

Inheritance Pattern

Myostatin-related muscle hypertrophy has a pattern of inheritance known as incomplete autosomal dominance. People with a mutation in both copies of the *MSTN* gene in each cell (homozygotes) have significantly increased muscle mass and strength. People with a mutation in one copy of the *MSTN* gene in each cell (heterozygotes) also have increased muscle bulk, but to a lesser degree.

Other Names for This Condition

- Muscle hypertrophy syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Myostatin-related muscle hypertrophy
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2931112/

Other Diagnosis and Management Resources

- GeneReview: Myostatin-Related Muscle Hypertrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1498

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Health Topic: Muscle Disorders
  https://medlineplus.gov/muscledisorders.html

Genetic and Rare Diseases Information Center

- Myostatin-related muscle hypertrophy
  https://rarediseases.info.nih.gov/diseases/10238/myostatin-related-muscle-hypertrophy

Educational Resources

- Disease InfoSearch: Myostatin-related muscle hypertrophy
  http://www.diseaseinfosearch.org/Myostatin-related+muscle+hypertrophy/5071
- MalaCards: myostatin-related muscle hypertrophy
  http://www.malacards.org/card/myostatin_related_muscle_hypertrophy
- Neuromuscular Disease Center, Washington University
  http://neuromuscular.wustl.edu/mother/mlarge.html#myostatinmut
- Orphanet: Myostatin-related muscle hypertrophy
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=275534

Patient Support and Advocacy Resources

- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/muscular.html

GeneReviews

- Myostatin-Related Muscle Hypertrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1498

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28myostatin+AND+muscle+hypertrophy%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- MYOSTATIN
  http://omim.org/entry/601788

Sources for This Summary

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- GeneReview: Myostatin-Related Muscle Hypertrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1498
- Schuelke M, Wagner KR, Stolz LE, Hübner C, Riebel T, Kömen W, Braun T, Tobin JF, Lee SJ. Myostatin mutation associated with gross muscle hypertrophy in a child. N Engl J Med. 2004 Jun 24;350(26):2682-8.
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